Case report

Weill- Marchesani Syndrome: a rare case report

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Abstract

Objective: To describe the presenting features of Weill-Marchesani syndrome

Case: A 22-year-old man presented with high myopia and progressive visual disturbance. He was of short stature and had brachydactyly. His initial Snellen best corrected visual acuity (BCVA) was 6/24 in his right eye and 6/12 in his left eye. Slit lamp examination revealed a sub-luxated micro-spherophakic lens. The patient was diagnosed with Weill-Marchesani syndrome. **Conclusion**: Weill-Marchesani syndrome can present wwith progressive myopia in a person of short stature and brachydactyly.

Key-words: Microspherophakia, brachydactyly, Weill-Marchesani syndrome

Introduction

Weill-Marchesani syndrome (WMS) is a rare connective tissue disorder first described by Weill in 1932, and further delineated by Marchesani (Marchesani et al 1939). Alternatively, it has been named spherophakia-brachymorphia syndrome or congenital mesodermal dysmorphodystrophy. Diagnositc criteria of WMS include short stature, brachydactyly and microspherophakia and/or ectopia lentis (Faivre et al, 2003). These patients may have joint stiffness and heart defects. Most patients have been described by ophthalmologists, since ocular symptoms and signs are characteristic of this syndrome and require clinical attention. Characteristic eye abnormalities consist of dislocation of the microspherophakic lens causing high myopia, acute and/or chronic angle-closure glaucoma, and cataracts. Despite the clinical homogeneity of disease presentation, autosomal recessive and autosomal dominant modes of inheritance have been reported (Dagoneau et al, 2004; Wirtz et al, 1996).

Knowledge of mode of presentation of this syndrome facilitates its timely diagnosis. We report a case of Weill-Marchesani syndrome to describe its presenting features.

Case report

A 22-year-old man presented with high myopia and progressive visual disturbance. His height was 136 cm (Fig 1 compares his height with the height of his brother, who is 7 years younger) and the body weight 43 kg. He had brachydactyly (Fig 2). His initial Snellen uncorrected visual acuity (UCVA) was 3/60 in the right eye and 4/60 in the left, and the best corrected visual acuity (BCVA) was 6/24 (-10 D sph/ -1 2.0 cyl at 35^0) in his right eye and 6/12 (-10.00 sph - 2.50 cyl at 140^0) in the left.

A-scan biometry revealed that the axial lengths of his eyeballs were 23.59 and 23.94 mm in the right and left eye respectively, suggesting a lenticular origin for the myopia. His intraocular pressure (IOP) was

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13 mmHg by Goldmann applanation tonometry in both eyes. Slit-lamp examination of both eyes before pupillary dilation showed normal corneas in both eyes. The anterior chambers were relatively shallow in the middle portion but normal at the periphery. There was bilateral anterior subluxation of the crystalline lens with the lens equator and zonules visible within the pupil (Fig 3). The iris around the pupillary border was pushed anteriorly which was prominently seen in undilated pupil. The posterior segment was normal in both eyes.

Figure 1: Showing short stature of the patient

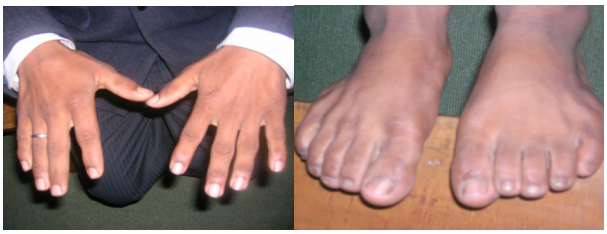

Figure 2: Brachydactyly

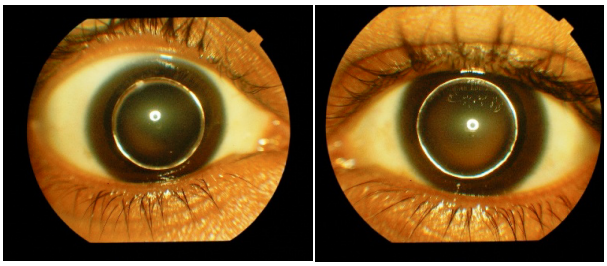

Figure 3: Anteriorly subluxated microspherophakic lens in both eyes.

Based on these findings, the patient was diagnosed with WMS. Since the patient showed improvement in visual acuity with refraction and the gonioscopy revealed that the angle structures were open, we did not persue active intervention. The patient was kept on regular follow up and advised to report back if he developed redness and pain.

Discussion

WMS is a rare connective tissue disorder characterized by short stature, brachydactyly, and spherophakia. Its mechanism is thought to be a developmental abnormality of the mesodermal origin tissues (ciliary body, lens, and epiphysis of bones). It is speculated that in spherophakia, the fetal lens which is physiologically spherical, is not subjected to the force of a properly acting ciliary body and zonules (Dietlein et al, 1996). Angle-closure glaucoma can occur in spherophakia through the pupillary block mechanism. This occurs when the dislocated lens moves into the anterior chamber which depends on the zonular integrity (Willi et al, 1973). Laser peripheral iridotomy can relieve pupillary block, and the area of appositional angle closure can be opened. In some cases, lens extraction may be needed.

Conclusion

Weill-Marchesani syndrome can present wwith progressive myopia in a person of short stature and brachydactyly.

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